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REVIEW ARTICLE NEWBORN SCREENING

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ABSTRACT

The first disorder detected by modern newborn screening programs was a metabolic condition in which the inability to degrade the essential amino acidphenylalanine can cause irreversible mental retardation unless detected early. With early detection, and dietary management, the negative effects of the disease can be largely eliminated. Robert Guthrie developed a simple method using a bacterial inhibition assay that could detect high levels of phenylalanine in blood shortly after a baby was born. Guthrie also pioneered the collection of blood on filter paper which could be easily transported, recognizing the need for a simple system if the screening was going to be done on a large scale. Newborn screening around the world is still done using similar filter paper.

KEYWORDS: Newborn Screening Program, Dry Blood Sample, Infant Screening Tests, Neonatal Screening Tests, The PKU Test

INTRODUCTION

Robert Guthrie is given much of the credit for pioneering the earliest screening for phenylketonuria in the late 1960s using blood samples on filter paper obtained by pricking a newborn baby's heel on the second day of life to get a few drops of blood.[1]Congenital hypothyroidism was the second disease widely added in the 1970s.[2] Guthrie and colleagues also developed bacterial inhibition assays for the detection of maple syrup urine disease and classic galactosemia.[3] The development of tandem mass spectrometry screening in the early 1990s led to a large expansion of potentially detectable congenital metabolic diseases that can be identified by characteristic patterns of amino acids and acylcarnitines.[4]

In the United States, the American College of Medical Genetics recommended a uniform panel of diseases that all infants born in every state should be screened for. They also developed an evidence-based review process for the addition of conditions in the future. The implementation of this panel across the United States meant all babies born would be screened for the same number of conditions. Prior to this, babies born in different states had received different levels of screening. On April 24, 2008, President George W. Bush signed into law the Newborn Screening Saves Lives Act of 2007. This act was enacted to increase awareness among parents, health professionals, and the public on testing newborns to identify certain disorders. It also sought to improve, expand, and enhance current newborn screening programs at the state level.

Wilson and Juegner Criteria for Disease Screening

- The condition sought should be an important health problem.
- There should be an accepted treatment for patients with recognized disease. Facilities for diagnosis and treatment

should be available.

- There should be a recognizable latent or early symptomatic stage.
- There should be a suitable test or examination.
- The test should be acceptable to the population.
- The natural history of the condition, including development from latent to declared disease, should be adequately understood.
- There should be an agreed policy on whom to treat as patients.
- The cost of case-finding (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole.
- Case-finding should be a continuing process and not a "once and for all project.

How is Newborn Screening Done?

Newborn Screening is done in Three Ways:

Most newborn screening is done with a blood test. Your baby's provider pricks your baby's heel to get a few drops of blood. The blood is collected on a special paper and sent to a lab for testing. The lab then sends the results back to your baby's health provider.

For the hearing screening, your provider places a tiny, soft speaker in your baby's ear to check how your baby responds to sound.

For heart screening, a test called pulse oximetry is used. This test checks the amount of oxygen in your baby's blood by using a sensor attached to his finger or foot. This test is used to screen babies for a heart condition called critical congenital heart disease (CCHD).

Why the Test is Performed

Screening tests do not diagnose illnesses. They identify which babies need additional testing to confirm or rule out illnesses. If follow-up testing confirms that the child has a disease, appropriate treatment can be started right away, before symptoms appear.

Screening tests are used to detect a number of disorders including: The health conditions are divided into five groups:

- **Organic Acid Metabolism Disorders**

Babies with these problems don't metabolize food correctly. Metabolism is the way your body changes food into the energy it needs to breathe, digest and grow.

Isovaleric acidemia (IVA)

Glutaric acidemia (GAI)

Hydroxymethylglutaric aciduria, also called 3-OH 3-CH3 glutaric aciduria (HMG)

Multiple carboxylase deficiency (MCD)

Methylmalonic acidemia, mutase deficiency (MUT)

3-methylcrotonyl-CoA carboxylase deficiency (3MCC)

Methylmalonic academia, CB1 A and CBl B forms (Cbl A,B)

Propionic acidemia (PROP)

Beta-ketothiolase deficiency (BKT)

- **Fatty Acid Oxidation Disorders**

When your body runs out of sugar, it usually breaks down fat for energy. A baby with fatty acid oxidation problems can't change fat into energy.

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

Carnitine uptake defect (CUD)

- **Amino Acid Metabolism Disorders**

Babies with these problems can't process amino acids in the body. Amino acids help the body make protein.

Phenylketonuria (PKU)

Maple syrup urine disease (MSUD)

Homocystinuria (HCY)

Citrullinemia (CIT)

Argininosuccinic acidemia (ASA)

Tyrosinemia type I (TYR I)

- **Hemoglobin Disorders**

These problems affect red blood cells. Red blood cells carry oxygen to the rest of the body.

Sickle cell anemia

Hb S/beta-thalassemia (Hb S/Th)

Hb S/C disease (Hb S/C)

- **Other Disorders**

Congenital hypothyroidism (HYPOTH)

Biotinidase deficiency (BIO)

Congenital adrenal hyperplasia (CAH)

Galactosemia (GALT)

Hearing loss (HEAR)

Cystic fibrosis (CF)

Severe combined immunodeficiency (SCID)

Critical congenital heart disease (CCHD)

How to Prepare for the Test

There is no preparation necessary for newborn screening tests. The tests are performed when the baby is between 24 hours and 7 days old, typically before the baby goes home from the hospital. The time of sampling is very essential. The sample must be taken on day 2 or before discharge, which comes first. Premature infants should be collected just before discharge, unless symptomatic.

Newborn Screening Program

All states have a Newborn Screening Program. The state's laws determine the disorders screened for by the program. The state program makes sure all babies in the state get screened. They also make sure that all babies with abnormal newborn screening test results are referred for follow-up care. Finally, they educate parents and doctors about newborn screening. Most newborn screening is done through state programs.

- **Blood Test**

Most newborn screening is done with a blood test. Your baby's provider pricks your baby's heel to get a few drops of blood. The blood is collected on a special paper and sent to a lab for testing. The lab then sends the results back to your baby's health provider.

Procedure for Blood Test

Position a sterile disposable lancet 9 2.0 – 2.4 mm tip) or an automatic lancet at a slight angle to perform a swift clean puncture

Wipe away the first drop of blood with dry sterile gauze.

Collecting dry Blood Sample;

Allow a second large drop of blood to form.

Lightly touch blood drop to soak through and completely fill circle.

Fill remaining circles with successive blood drops.

Do Not

Milked or squeezed the baby's heel to obtain the blood, just leave to flow in appropriate drop size

Used syringes or needles to aspirate blood for collecting blood spot, as this may cause oversaturation of blood on the filter paper or scratch it.

How to Dry DBS

DBS must be air dried for at least 3-4 hours at room temperature

Cards can be dried in a drying rack

All cards must be totally dried before sending to the lab.

Safety first Be Careful;

During the drying process;

Avoid touching or smearing the blood spots

Keep it away from direct sunlight

Should not be dried near an open window, avoid dust and flying insects

- **Hearing Screen**

Two different tests can be used to screen for hearing loss in babies. Both tests are quick safe and comfortable with no activity required from your child, these tests are often performed while the baby is asleep.

- **Otoacoustic Emission Test OAE**

This test is used to determine certain parts of baby's ears respond to sound. During the test a miniature earphone and microphone are placed in the ear and sounds are played. When a baby has normal hearing, an echo is reflected back into the ear canal, which can be measured by the microphone. If no echo is detected, it can indicate hearing loss.

- **Auditory brain stem response Test ABR**

This test is used to evaluate the auditory brain stem (the part of the nerve that carries sound from the ear to the brain stem) and the brains response to sound. During the test a miniature earphone and microphone are placed in the ear and sounds are played. Band-Aid- like electrodes are placed along the baby's head to detect the brains response. If the baby's brain doesn't respond consistently to the sounds there may be a hearing problem.

- **Heart Screen - Pulse Oximetry Testing**

Pulse oximetry is a non invasive test that measures how much oxygen in the blood. Baby with heart problem may have low blood oxygen level; therefore it can help identify babies that may have critical congenital heart disease. This test is done by a machine called pulse oximeter.

Every 15 minutes a baby is born with a congenital heart defect and much attention has been given to the early detection of this serious condition in newborns. In 2011 , the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children as well as the American Academy of Pediatrics, The American college of Cardiology, and the American Heart Association have come together to recommend the use of pulse oximetry to detect critical congenital heart disease during a newborn screening.

While many states are at various points of legislative position, most hospitals and nurseries are evaluating the implementation of pulse oximetry screening as a part of their newborn assessment plan.

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